Letter to the Editor

Further Case of Galloway-Mowat Syndrome of Abnormal Gyral Patterns and Glomerulopathy

To the Editor:

We read with great interest the article by Cooperstone et al. [1993] in this Journal reporting 3 new cases of Galloway-Mowat syndrome of abnormal gyral patterns and glomerulopathy. We wish to add another case to the literature.

CLINICAL REPORT

The patient was a male Caucasian infant born to a healthy 38-year-old gr 2 ab 1 mother in the 36th week of gestation. The father was 36 years old and in good health. There was no consanguinity. Amniocentesis performed at 16 weeks gestation for advanced maternal age demonstrated a normal male karyotype (46, XY). Antenatal ultrasound study at this time did not demonstrate any abnormality. The pregnancy had been uncomplicated. Birth was by spontaneous vaginal delivery. The Apgar scores were 6 and 9 at 1 and 5 minutes, respectively. Birth weight was 2,278 g (50th centile), length 50 cm (50th centile), and head circumference (OFC) 28 cm (<10th centile). The placenta was normal.

The baby was stable, but in the second week of life he developed edema, poor feeding and regurgitation. He was referred at 10 days for investigation. At that time he had microcephaly, micrognathia, high arched palate, apparently low-set posteriorly angulated ears, overlapping fingers, flexion deformity of the thumbs, bilateral simian creases, widely spaced nipples, inguinal testes, marked pitting pedal edema, ballottable kidneys, poor muscle tone and decreased spontaneous movements.

Results of biochemical investigation: elevated blood urea (11.4 mmol/L) and creatinine (92 mmol/L) and low serum protein (17 g/L) which was associated with an increased urinary protein excretion (81 g/L). No evidence for toxoplasmosis, rubella, cytomegalovirus, herpes, and syphilis infections was found.

Renal ultrasound scan demonstrated diffusely increased echogenicity with poor corticomedullary differentiation, and increased length of kidneys for gestation (5.3 cm). Cranial ultrasound study demonstrated a smooth simple contour of both cerebral hemispheres with lack of normal sulcal pattern, suggestive of pachygyria. A CT scan showed an absence of sulci and gyri and reduced density of the white matter. Upper gastrointestinal contrast studies demonstrated a large fixed hiatus hernia, marked gastro-esophageal reflux and esophageal dysmotility. Although no overt seizures were seen or reported the EEG was severely abnormal with moderately to severely suppressed background and frequent, almost persistent, unusual theta bursts originating from both central regions of the brain while active and during quiet sleep.

His general condition deteriorated with increasing oedema, renal failure and hypoalbuminaemia. He died at age 32 days; autopsy demonstrated a small brain, weight only 172 g (expected wt. 431 ± 117.7 g), with a smooth appearance of the frontal lobes. Microscopically there was fusion of the leptomeninges and molecular layer, heterotopic neurons, astrocytosis, abnormal or failed lamination of the cortex, dysplasia of the dentate nucleus, and frontal pachygyria. Renal examination demonstrated hyperlobulation of the kidneys and, on microscopy, uniformly small glomeruli, reduced capillary loop formation and generalised sclerosis. Also, there was a fixed hiatus hernia, thyroid dysplasia and adrenal hypoplasia, marked edema, pleural and pericardial effusions.

The present case adds to the number of cases of Galloway-Mowat syndrome. The diagnosis was made on the basis of microcephaly, congenital nephrosis and hiatus hernia. Most of the findings in this case have been described before, whereas thyroid dysplasia and adrenal hypoplasia are likely components of the syndrome, but not previously described.

REFERENCES

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